

Human Biological Sex and Intersex Variations

Compiled by Dan Hagler for Educational and Reference Use

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Condition	Chromosomes	Gonads	Hormones	External Genitalia / Traits
Cisgender Male	46,XY	Testes	Typical male (testosterone)	Male anatomy
Cisgender Female	46,XX	Ovaries	Typical female (estrogen)	Female anatomy
Klinefelter Syndrome	47,XXY	Small testes	Low testosterone	Male, breast development possible, infertile
Turner Syndrome	45,X or 45,X0	Streak gonads	Low estrogen	Female, short stature, infertile
Triple X Syndrome	47,XXX	Ovaries	Normal	Female, usually typical phenotype
XYY Syndrome	47,XYY	Testes	Normal to elevated T	Male, tall stature, may have learning issues
Complete AIS	46,XY	Undescended testes	Normal T, but ineffective	Female appearance, no uterus, infertile
Partial AIS	46,XY	Testes	Partial androgen response	Ambiguous / partially male or female traits
5-Alpha-Reductase Deficiency	46,XY	Testes	Low DHT conversion	Female/ambiguous at birth, masculinizes at puberty
Congenital Adrenal Hyperplasia	46,XX or 46,XY	Ovaries (XX) / Testes (XY)	Excess androgens (in XX)	XX may have virilized genitalia; XY usually typical
Ovotesticular DSD	46,XX / 46,XY / Mosaic	Ovary + testis or ovotestis	Varies	Ambiguous or mixed features
Mixed Gonadal Dysgenesis	45,X/46,XY mosaic	Testis + streak gonad	Varies	Ambiguous; often male-leaning with atypical traits
Mosaicism / Chimerism	XX/XY combinations	Mixed or dual types	Varies	Wide range of outcomes
De la Chapelle Syndrome	46,XX with SRY	Testes	Testosterone present	Male appearance, infertile
Swyer Syndrome	46,XY	Streak gonads	Low sex hormones	Female external traits, no puberty without HRT